



## Overview

### Overview of Pachyonychia Congenita

Pachyonychia congenita (PC) is a very rare genetic disorder that affects the skin and nails. The symptoms usually begin at birth or early in life, and the condition affects people of both sexes and all racial and ethnic groups.

PC is caused by mutations affecting keratins, proteins that provide structural support to cells, and it is classified into five types based on which keratin gene harbors the mutation. Symptoms vary from person to person and depend on the type, but thickened nails and calluses on the soles of the feet occur in almost all cases. The most debilitating symptom is painful calluses on the soles that make walking difficult. Some patients rely on a cane, crutches, or a wheelchair to help manage the pain of walking.

There is no specific treatment for PC, but there are ways to manage the symptoms, including the pain.

## Who Gets

### Who Gets Pachyonychia Congenita?

People who have pachyonychia congenita have a mutation in one of five keratin genes. Researchers have found more than 115 mutations in these genes that are linked to the disorder. In some cases, PC is inherited from a parent, while in others, there is no family history and the cause is a spontaneous mutation. The disorder is genetically dominant, which means that a single mutated gene copy is enough to cause the condition. PC is very rare. It affects people of both sexes and all racial and ethnic groups.

## Types

### Types of Pachyonychia Congenita

There are five types of pachyonychia congenita, and they are classified based on the keratin gene that is altered. Thickened nails and painful calluses on the soles of the feet are typical of all forms of the disorder, but the presence of other features can depend on which keratin gene is affected, and possibly on the specific mutation.

## Symptoms

### Symptoms of Pachyonychia Congenita

The symptoms and severity of PC can vary widely, even among people with the same type or in the same family. Most symptoms typically appear within the first months or years of life.

The most common features of PC include:

- **Painful calluses and blisters** on the soles of the feet. In some cases, the calluses itch. Calluses and blisters may also form on the palms of the hands.
- **Thickened nails.** Not all nails are affected in every patient with PC, and some people do not have any thickened nails. But the vast majority of patients have some affected nails.
- **Cysts** of various types.
- **Bumps around hairs at friction sites**, such as the waist, hips, knees, and elbows. They are most common in children and lessen after the teenage years.
- **White film on the tongue and inside the cheeks.**

Less common features of PC include:

- **Sores at the corners of the mouth.**
- **Teeth at or before birth.**
- **White film on the throat**, resulting in a hoarse voice.
- **Intense pain on first bite** (“first bite syndrome”). The pain is near the jaw or ears and lasts 15–25 seconds when beginning to eat or swallow. This is more common in younger children and may cause feeding difficulties for some infants. It typically goes away during the teenage years.

## Causes

### Causes of Pachyonychia Congenita

Pachyonychia congenita is caused by mutations in genes that encode keratins, proteins that are the main structural components of skin, nails, and hair. The mutations prevent keratins from forming the strong network of filaments that normally gives skin cells strength and resilience. As a result, even normal activities like walking can cause cells to break down, ultimately leading to the painful blisters and calluses that are the most debilitating features of the disorder.

## Diagnosis

### Diagnosis of Pachyonychia Congenita

Doctors usually diagnose PC by:

- Completing a physical exam, including examination of the skin and nails.
- Asking about the family and medical history, as many cases of PC are inherited.
- Ordering a genetic test. By identifying the disease mutation, a genetic test can rule out other conditions with similar symptoms and confirm that a person has PC. A genetic test may also provide a clearer picture of what to expect, as symptoms of PC differ based on the mutation.

## Treatment

### Treatment of Pachyonychia Congenita

There is no cure and there are no specific therapies for PC. The main goal of treatment is relieving the pain by most people with the condition.

Your doctor may recommend the following treatments for calluses:

- **Thinning the calluses.** Regular grooming of the feet is a mainstay of treatment. It is important not to trim too aggressively, as overly thinning the calluses can increase the pain. Conversely, trimming regularly, as needed, is important because overly thick calluses can also increase the pain.
  - Physical abrasion, such as by paring or filing the callused areas, is the most common approach. Soaking the skin prior to abrading is helpful for some people. While many people pare or file their calluses themselves, others see a podiatrist, a medical professional who specializes in caring for the feet.
  - Oral retinoids, which are related to vitamin A, can thin calluses, but they may not decrease the pain, and in some cases they increase it. If a doctor prescribes these, he or she will carefully monitor the dosage and duration of treatment.
- **Moisturizing creams or lotions.** Moisturizers can provide relief by softening the skin and preventing cracks.

- **Over-the-counter or prescribed anti-inflammatory and pain medications.** These may help temporarily alleviate pain and are sometimes taken prior to engaging in physical activity to “get ahead” of the anticipated pain.
- **Special orthotics or insoles.** These can redistribute the weight on your feet and provide relief from pain. In severe cases, a person may need a cane, crutches, or a wheelchair.
- **Nails and cysts.** Your doctor may recommend the following treatments for thickened nails and cysts:
  - **Thickened nails.**
    - Regular trimming. Nails are not usually painful, but they can become so if they become infected or broken.
    - Bleach baths. Routinely bathing your nails and feet in mild bleach solutions can help prevent infections. You should not use this treatment without first talking to your doctor.
    - Oral antibiotic or antifungal medications. These may be needed if infections develop.
    - Surgery to remove especially troublesome nails.
- **Cysts.** These may need to be drained or surgically removed. Your doctor may prescribe antibiotics if cysts become infected. Antiseptic cleansers may help prevent flare-ups.

## Who Treats

### Who Treats Pachyonychia Congenita?

Dermatologists, who specialize in skin disorders, usually treat PC. Other health care professionals who may be involved in your care include:

- Clinical geneticists, who diagnose and treat children and adults with genetic disorders.
- Mental health professionals, who can help you cope with difficulties in the home and workplace that may result from having the disorder.
- Podiatrists, medical specialists who provide care for the feet and lower legs.
- Primary care doctors, such as family physicians, internal medicine specialists, and pediatricians, who coordinate care between the different health providers and treat other problems as they arise.

## Living With

### Living With Pachyonychia Congenita

Having a painful disorder like PC can be challenging, but the following may make it easier to manage:

- Maintain a healthy weight, and limit walking and standing. This may lessen the pain from the calluses on the soles of the feet.
- Wear comfortable shoes and socks that reduce moisture. This may help decrease rubbing that can worsen painful calluses.
- Wear gloves to protect the hands during activities like riding a bicycle or using hand tools.
- If you have white patches in the mouth, brush your teeth and tongue frequently and gently to reduce their appearance. Infants with this symptom may feed better if you use a bottle with a soft nipple and a wide opening.
- Use moisturizers, such as those that contain petroleum jelly or lanolin, to soothe bumps that develop near friction sites, such as the waist, elbows, and knees.
- Visit a mental health professional or join a support group. These can provide emotional support and help you cope with feelings of isolation that often come with having a rare disorder.

Remember to visit your health care providers regularly and to follow their recommendations.

## Research Progress

### Research Progress Related to Pachyonychia Congenita

Investigators at the National Institutes of Health and other research centers across the country are working to develop therapies for PC. Much of the research focuses on targeting the underlying source of the disorder by “shutting down” the mutated keratin gene. Other efforts are aimed at reducing the pain associated with the symptoms. Following are examples of ongoing research studies.

- **Targeting the mutated genes.**
  - Investigators have uncovered evidence that a class of approved medications can improve symptoms of PC. Researchers are exploring the mechanisms of the drugs’ action, which likely includes lowering cellular levels of the mutated keratins, and are testing their benefit in a clinical trial of people with PC.
  - Small interfering RNA (siRNA). Scientists are using siRNA, a genetic tool for silencing specific genes, to target the altered keratin genes in people with the disorder.
- **Alleviating pain.**
  - The cause of pain in PC is poorly understood, but some evidence suggests that neuropathic pain, which stems from damaged or injured nerves, is at least partly to blame.

Studies are testing the effectiveness of drugs that treat this type of pain.

- **Characterizing each type of the condition.**

- Researchers are working to map the features, severity, and course of each type of PC.

With a clearer picture of the characteristics of each type, they will more easily be able to design clinical trials to accurately measure the efficacy of experimental therapies.

## Related Resources

### For More Info

#### **U.S. Food and Drug Administration**

Toll free: 888-INFO-FDA (888-463-6332)

Website: <https://www.fda.gov>

Drugs@FDA at <https://www.accessdata.fda.gov/scripts/cder/daf>. Drugs@FDA is a searchable catalog of FDA-approved drug products.

#### **Centers for Disease Control and Prevention, National Center for Health Statistics**

Website: <https://www.cdc.gov/nchs>

#### **NIH Office of Rare Diseases Research Genetic and Rare Diseases Information Center**

Website: <https://rarediseases.info.nih.gov>

#### **National Library of Medicine's Genetics Home**

##### **Reference**

Website: <https://ghr.nlm.nih.gov/condition/pachyonychia-congenita>

#### **Pachyonychia Congenita Project**

Website: <https://www.pachyonychia.org>

#### **American Academy of Dermatology**

Website: <https://www.aad.org>

Si necesita más información sobre recursos confiables en su idioma o en otro idioma, por favor visite las páginas web que aparecen a continuación o comuníquese con el Centro Nacional de

Distribución de Información del NIAMS en NIAMSInfo@mail.nih.gov.

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